APPROACH TO DEVELOPMENTAL DELAY

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Definition:

- Delay in the acquisition of developmental skills beyond limit age.

Types:
- Isolated
- Global
ISOLATED DELAY

- Gross motor delay
- Impaired speech and language
- Impaired hearing
- Visual impairment
- Impaired cognition/learning difficulties
- Impaired social skills / Autism
GLOBAL DELAY

- Delay in acquisition of two or more skill fields.
- Presents within first two years of life
CAUSES:

- Gross motor delay:
  - Cerebral palsy
  - Neuromuscular disorders
Static Global delay:

- Chromosomal disturbances
- Maternal alcohol or drug abuse
- Cerebral malformations
- Hypoxic ischemic encephalopathy
- Intracranial hemorrhage
- Infection
- Brain tumor
Progressive global delay:

1. NEUROCUTANEOUS DISORDERS
   Tuberous sclerosis
   Neurofibromatosis
2. METABOLIC BRAIN DISORDERS

A. Gray matter disorders (without visceral storage)
   - Tay Sachs disease
   - Lipofuscinosis
   - Alpers disease
   - Menkes disease
   - Rett syndrome
B. Gray matter diseases (with visceral storage)
GM-1 gangliosidosis
GM-2 gangliosidosis (Sandhoff)
Niemann-Pick disease
Gaucher disease
C. Diseases affecting white matter:
   Galactosemia
   Phenylketonuria
   Maple syrup urine disease
   Homocystinuria
   Urea cycle defects
D. Diseases affecting white & gray matter
MPS I, II, III, VII
Mitochondrial disorders
Peroxisomal disorders
3. Congenital hypothyroidism
4. Malnutrition
5. Chronic illness
CEREBRAL PALSY

- Disorder of tone and posture due to nonprogressive lesion of the developing brain
- Often associated with impaired speech, hearing and cognition.
- Birth history: asphyxia, prematurity
- Developmental delay
- Seizures
- Microcephaly
- Quadriplegia
- Paraplegia
- Hemiplegia
- Diplegia
- Spasticity
- Hypotonia & ataxia
- Dyskinetic
Neuromuscular disorders:

- Anterior horn, nerves, NMJ & muscle
- SMA
- Myotonias
- Myopathies
- Mysthenia gravis
Chromosomal disturbances

Most common---trisomy 21
Typical facies
Hypotonia
Congenital heart disease
Karyotyping
- Edward syndrome (trisomy 18)
- Developmental delay
- Rocker bottom feet
- Finger overlap
- Fragile X syndrome
- Prominent forehead
- Large ears
- Macrocephaly
- Large testes
- Karyotyping
Hypoxic ischemic encephalopathy

- Motor, cognition, seizures
- Perinatal asphyxia
- Cardiopulmonary disease
- Meningitis
- Head trauma
- Status epilepticus
- CT/MRI
Maternal drugs/alcohol

- Alcohol ---- fetal alcohol syndrome
- Cocaine --- cerebral hemorrhage & infarction
- Anticonvulsants ---- cerebral malformations & dev delay
FETAL ALCOHOL SYNDROME:
  microcephaly, thin upper lip, flattened midface.
  brain dysfunction, developmental delay, poor memory
Congenital infections

- Toxoplasmosis
- Rubella
- CMV
- Herpes simplex
- Syphilis
Growth restriction

Eye defects: cataracts, microphthalmia, retinitis

Pneumonitis

Hepatomegaly, Jaundice, Hepatitis

Virus in urine

Bone abnormalities

Intracerebral calcification, Hydrocephalus

Microcephalus, Deafness

Heart defects: cardiomegaly, patent ductus arteriosus

Splenomegaly

Rash: blueberry muffin or petechial

Anaemia, Neutropenia, Thrombocytopenia

Brain infections

- Bacterial / tuberculous
- Herpes encephalitis
- Seizures
- Motor & cognitive impairment
- Language dysfunction
- Deafness & blindness.
Neurocutaneous Syndromes

- Neurofibromatosis
  - Autosomal dominant…1 in 3000
  - Diagnostic criteria (2 or more)
    - 6 or more café-au-lait spots >5mm
    - >1 neurofibroma
    - Axillary freckles
- Optic glioma
- Lisch nodule of iris
- Sphenoid dysplasia
- First degree relative with NF1.
Tuberous sclerosis

- Autosomal dominant.......1 in 9000
- Cutaneous features:
  ash leaf patches(depigmented)
  shagreen patches(roughened skin)
  adenoma sebaceum on face
  (angiofibromata)
Sturge webber syndrome

- Port wine stain
- Seizures
- Learning disability
- Hemiplegia
GM2 Gangliosidosis (Tay-Sachs disease)

- Autosomal recessive
- Deficient Hexosaminidase
- Onset 3 – 6 mo
- Abnormal startle reaction
- Developmental delay
- Cherry red spot
- Enzyme analysis in cultured leukocytes
Mucopolysaccharidosis (I, II, III, VII)

- Lysosomal enzymes deficiency
- AR (except II...X linked)
- Coarse facial features,
- Hepatosplenomegaly
- Dysostosis multiplex
- Corneal clouding (except II&III)
Congenital hypothyroidism

- Developmental delay
- Mental retardation
- Coarse features
- Neonatal jaundice
- Constipation
- Umbilical hernia
Diagnostic approach
HISTORY

- Antenatal history (maternal infections, cardiovascular diseases, vaccination)
- Birth history
- Prematurity
- Fever, jaundice
- Feeding
FAMILY HISTORY:
- consanguinity
- other siblings
- developmental delay
- death of siblings
● Developmental history: milestones achieved & at which ages. unAchieved milestones
● Chronic complaints
  fever
  cough
  vomiting
  diarrhea
  recurrent ARI
  cyanosis & fatigability

● Seizures
EXAMINATION

- Anthropometry
- Developmental assessment (Denver dev. Screening test)
  hearing, vision, fine & gross motor, primitive reflexes.
● Neurological examination
● Abdominal examination
● Eye examination
  shape, opacities, cataracts, cherry red spot.
● Neurocutaneous stigmata
Developmental Delay

- speech
- motor
- global
- Delayed Speech
- hearing loss
- autism
- pseudobulbar palsy
GLOBAL DELAY

DYSMORPHISM

H/O birth asphyxia, infection, trauma, drugs

CP

YES

CT/MRI
DYSMORPHIC

CT/MRI

NO

MALFORMATIONS

Chromosomal analysis
CHROMOSOMAL ANALYSIS

YES                                    NO
SYNDROMES                    TORCH SCREEN
VISCEROMEGALY

NO

CUTANEOUS STIGMATA

YES

COARSE FACIES

T4/TSH
VISCEROMEGALY

COARSE FACIES

NO

REDUCING SUBSTANCES IN URINE

YES

MUCOPOLYSAC-CHRIDURIA
REDUCING SUBS IN URINE

NO

GAUCHER CELLS

YES

GAUCHER DISEASE

NO

ENZYME ANALYSIS

YES

GALACTOSEMIA
Other tests

- Audiologic testing
- Ophthalmologic evaluation
- EEG
- EMG, Nerve conduction studies, muscle biopsy
THANK YOU.